

(19) World Intellectual Property
Organization
International Bureau



(43) International Publication Date
6 May 2005 (06.05.2005)

PCT

(10) International Publication Number
WO 2005/040335 A2

- (51) International Patent Classification⁷: C12N (74) Agent: ALBIHNS STOCKHOLM AB; Box 5581, S-114 85 Stockholm (SE).
- (21) International Application Number: PCT/SE2004/001549 (81) Designated States (unless otherwise indicated, for every kind of national protection available): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NA, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.
- (22) International Filing Date: 25 October 2004 (25.10.2004)
- (25) Filing Language: English
- (26) Publication Language: English
- (30) Priority Data:
0302789-3 23 October 2003 (23.10.2003) SE
60/481,546 23 October 2003 (23.10.2003) US
- (71) Applicants and
(72) Inventors: **HOLMBERG, Monika** [SE/SE]; Dep. of Biosciences, Unit. of Clinical and Medical Genetics, Umeå University, S-901 87 Umeå (SE). **HOLMBERG, Dan** [SE/SE]; Dep. of Biosciences, Unit of Clinical and Medical Genetics, Umeå University, S-901 87 Umeå. **MINDE, Jan** [SE/SE]; Dep. of Orthopaedics, Gällivare Hospital, S-982 82 Gällivare (SE). **HOLMGREN, Gösta** [SE/SE]; Dep. of Biosciences, Unit of Clinical and Medical Genetics, Umeå University, S-901 87 Umeå (SE). **TOOLANEN, Göran** [SE/SE]; Dep. of Surgery and Perioperative Sciences, Unit of Orthopaedics, Umeå University, S-901 85 Umeå (SE).
- (84) Designated States (unless otherwise indicated, for every kind of regional protection available): ARIPO (BW, GH, GM, KE, LS, MW, MZ, NA, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PL, PT, RO, SE, SI, SK, TR), OAPI (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).
- Published:**
— without international search report and to be republished upon receipt of that report
- For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

(54) Title: A NOVEL MUTATED NERVE GROWTH FACTOR BETA GENE, PROTEINS ENCODED THEREBY, AND PRODUCTS AND METHODS RELATED THERETO

(57) Abstract: A point mutation responsible for loss of pain perception is identified and disclosed. The loss of pain perception resulting from the genetic mutation is characterized by impairment in the sensing of deep pain and temperature but with mental abilities and most other neurological responses intact. The mutation occurs in the coding region of the nerve growth-factor beta (NGFB). The NGF mutation separates the effect of NGF mediating a normal development of functions of the central nervous system, such as mental abilities, from effects on the nervous system involved in peripheral pain pathways. New screening methods, research tools, and therapies involving pain pathways related to the point mutation are provided.



WO 2005/040335 A2